A rare form of Turner Syndrome diagnosed in the first trimester.

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Introduction
Monosomy X is present in about 3% of all conception. The usual ultrasound findings are: nuchal cystic hygroma, horseshoe kidney, left-sided cardiac anomalies, nonimmune fetal hydrops. The most common cardiac defects are bicuspid aortic valve and coarctation of the aorta.

Case report
A 31 years old primigravida, 14 weeks of amenorrhea, without any history of drugs, alcohol and tobacco use or any known diseases was examined for the first trimester screening.

The transabdominal ultrasound revealed: a fetus with a CRL-73.5 mm, FHR-155 bpm, amniotic fluid in normal amount, normal looking placenta, a 3-vessel cord, normal extremities nuchal translucency-1.7 mm, present nasal bone and a ductus venosus PI-3.05 with a negative A wave. We proceeded to transvaginal ultrasound for the better assessment of the heart. We found a tricuspid atresia type 1 with a vsd and a normal aortic arch.

Maternal serum biochemistry
Free bHCG-0.4 MoM; PAPP-A-0.54 MoM

The combined calculated risk result
T18 1:88, T13 1:692, T21 1:245

Karyotype result
The couple decided to terminate the pregnancy and underwent genetic investigation that revealed 45XO/46XY mosaicism. Furthermore, the karyotype of the couple revealed polymorphism of the 9th chromosome, for both of the parents.

Conclusion
Cardiac anomalies can be diagnosed in the first trimester using a transvaginal probe for better visualisation. In our case the cardiac anomalies were the main trigger for abnormal karyotype.